(FILE 'HOME' ENTERED AT 17:12:50 ON 22 NOV 2002)

FILE 'MEDLINE, CAPLUS' ENTERED AT 17:12:59 ON 22 NOV 2002
L1 49 S CD38 (P) GENE (P) SEQUENCE
L2 30 DUP REM L1 (19 DUPLICATES REMOVED)

FILE 'STNGUIDE' ENTERED AT 17:20:25 ON 22 NOV 2002

FILE 'MEDLINE, CAPLUS' ENTERED AT 17:25:42 ON 22 NOV 2002
L3 10 S CD38 AND (DIABETES OR IDDM OR NIDDM) AND (MUTATION? OR POLYMO
L4 7 DUP REM L3 (3 DUPLICATES REMOVED)
L5 6 S L4 NOT L2

FILE 'STNGUIDE' ENTERED AT 17:27:33 ON 22 NOV 2002

FILE 'MEDLINE, CAPLUS' ENTERED AT 17:34:56 ON 22 NOV 2002
L6 17 S CD38 AND INSULIN AND (MUTATION? OR POLYMORPHISM?)
L7 13 DUP REM L6 (4 DUPLICATES REMOVED)
L8 6 S L7 NOT L4

FILE 'STNGUIDE' ENTERED AT 17:37:12 ON 22 NOV 2002

FILE 'MEDLINE, CAPLUS' ENTERED AT 17:39:27 ON 22 NOV 2002
L9 267 S (MODY1 OR MODY2 OR MODY3) AND (MUTATION? OR POLYMORPHISM?)
L10 171 DUP REM L9 (96 DUPLICATES REMOVED)
L11 6 S L10 AND TYPE 1 AND TYPE 2

L11 ANSWER 3 OF 6 MEDLINE
AN 2001322865 MEDLINE
DN 21202573 PubMed ID: 11307309
TI Diabetes mellitus.
AU Iwasaki N
CS Diabetes Center, Tokyo Women's Medical University, Shinjyuku-ku, Tokyo 162-8666.
SO RINSHO BYORI. JAPANESE JOURNAL OF CLINICAL PATHOLOGY, (2001 Feb) 49 (2) 161-4. Ref: 0
Journal code: 2984781R. ISSN: 0047-1860.
CY Japan
DT Journal; Article; (JOURNAL ARTICLE)
General Review; (REVIEW)
(REVIEW LITERATURE)
LA Japanese
FS Priority Journals
EM 200106
ED Entered STN: 20010611
Last Updated on STN: 20010611
Entered Medline: 20010607
AB Diabetes mellitus is a group of metabolic disorders characterized by hyperglycemia resulting from defects in insulin secretion, insulin action or both. Genetic factors contribute to the development of diabetes. Some forms such as the condition called maturity-onset diabetes of the young(MODY) result from **mutations** in a single gene. Other forms such as **type 1** or **type 2** diabetes
are multifactorial in origin with different combinations of genes together with non-genetic factors contributing to the development of hyperglycemia. MODY has been a good model for studying the genetics and pathophysiology of diabetes. This form of diabetes can result from **mutations** in
at least seven different genes: hepatocyte nuclear factor(HNF)-4 alpha/**MODY1**, glucokinase/**MODY2**, HNF-1 alpha/**MODY3**,
insulin promoter factor(IPF-1)/MODY4, HNF-1 beta/MODY5, NeuroD1/MODY6 and Islet(Isl)-1/MODY7. **Mutations** in HNF-1 alpha/**MODY3** are
the most common cause of MODY in Japanese identified to date accounting for about 15% of cases of MODY. **Mutations** in the HNF-4 alpha/**MODY1**, glucokinase/**MODY2**, HNF-1 beta/MODY5 and
Isl-1/MODY7 genes have also been found in Japanese; however, they are rare causes of MODY. Clinical studies indicate that patients with MODY are generally not obese and that all forms of MODY are characterized by pancreatic beta-cell dysfunction. Patients who have **mutations** in the HNF-1 beta/MODY5 gene have non-diabetic kidney dysfunction including renal cysts. Female carriers may also exhibit abnormalities in the upper vagina and uterus. Genetic approach for **type 2**
diabetes had done by using non-parameteric linkage analysis such as sibpair analysis which worked well and NIDDM1 and NIDDM2 have been identified to date. The responsible gene for NIDDM1 was recently identified to be Calpain 10, and SNP43 in this gene could explain all of the evidence for linkage in Mexican American **type 2**
diabetes.

L2 ANSWER 10 OF 30 CAPLUS COPYRIGHT 2002 ACS
AN 2000:815089 CAPLUS
DN 133:359765
TI Diabetes risk factor detection with CD38 gene mutations
IN Egashira, Toru; Nagano, Makoto; Sagehashi, Yukiko; Matsui, Kana; Hattori, Hiroaki; Kanezuka, Akira; Takasawa, Noboru; Okamoto, Hiroshi
PA BML Inc., Japan
SO Jpn. Kokai Tokkyo Koho, 19 pp.
CODEN: JKXXAF
DT Patent
LA Japanese
FAN.CNT 1

	PATENT NO.	KIND	DATE	APPLICATION NO.	DATE
PI	JP 2000316578	A2	20001121	JP 1999-131955	19990512

AB Assessment of diabetes risk factors by detecting mutations in gene coding for type II transmembrane glycoprotein CD38 (ADP-ribosyl cyclase/cyclic ADP-ribose hydrolase), is disclosed. Polymorphism was obsd. in the exon 3 and 4 of CD38 gene from peripheral leukocytes; the corresponding point mutation of exon 3 show R140W missense mutation and that of exon 4 showed silent mutation at I 168. The allele frequency of R140W missense mutation was different between NIDDM and non-diabetic controls. Glut 2 gene mutation was also obsd. in a family with the CD38 gene mutation, indicating the involvement of CD38 gene and other genes in pathogenesis of NIDDM. DEEG (denaturing gradient gel electrophoresis), direct sequencing, and PCR-RFLP were used.